TITLE: KROMATID 5-Color Whole Genome ANALYSIS REPORT

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I. PROJECT INFORMATION

Project Quote #	
Sample Type	Whole Blood, cell type, etc
Sample ID	LIMS ID, customer sample name
Gender	Male
Passage number (or N/A)	N/A
Study Objective	

II. CHROMOSOME HARVEST

Harvest is performed per SOP-0044.

Condition	Result
Analog incubation time:	**24 hours**
Colcemid incubation time	**1 or 2 hours, both tested successfully**
Harvest Option	**Standard Protocol: X** **Modified Conditions: N/A**

III. dGH SCREEN ASSAY

Assay	Standard dGH SCREEN **AS-0002.1 (50 Cells)**
Metaphase and Karyotype Qualification	Samples must be prepared and qualified for dGH analysis prior to running the assay. dGH SCREEN is designed for samples with grossly normal karyotypes and has not been qualified for highly rearranged genome analysis. Spread resolution of 550+ (G-band equivalent) is recommended.
Assay Description	The five-color whole genome assay (5CWG or dGH SCREEN) is a dGH paint combination assay for all 24 human chromosomes. The assay is composed of standard design, high-density (HD) dGH chromosome paints in five color panels such that chromosomes painted in the same color can be differentiated by size, shape, and centromere position. Results include per-chromosome attribution of inter and intra-chromosomal structural events including inversions, translocations, aneuploidy (gain and loss), insertions, centromere abnormalities and complex events across a sample. Prior to analysis, images of dGH SCREEN painted metaphase spreads are qualified, processed and sorted into karyograms for rapid, consistent reading of the assay. Per-cell event assessment is performed in an excel workbook which is built to be leveraged for population-level analysis of events ranging from random to clonal.

IV. RESULTS

Cells Analyzed	50	Total Karyograms	50

Chromosome Aberration Data:

Table 1: Structural Event Summary

Event	Number of Events	Number Of Cells with Events
Gain	4	4
Loss	1	1
Inversion	110	39
SCE	225	50
Insertion	0	0
Size Diff	1	1
Translocations	5	3
Complex	1	1
Total Events per Chromosome	**347**	**99**

Table 2: Distribution of Events by Chromosome

Count of Events by Chromosome																									
Chromosome	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	X	Y	Num of Events	Num of Cells w/ Events
Gain	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	4	0	4	4
Loss	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	1	0	1	1
Inversion	9	6	3	5	4	11	3	7	12	1	2	5	4	11	4	6	0	7	1	2	0	3	4	110	39
SCE	16	18	26	17	18	8	12	12	9	14	9	8	12	6	6	6	6	9	2	1	2	1	7	225	50
Insertion	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
Size Diff	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	1	1
Translocations	1	1	0	0	0	0	0	2	0	0	0	0	0	1	0	0	0	0	0	0	0	0	0	5	3
Complex	0	0	0	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	1	1
Total Events per Chromosome	26	25	30	22	22	20	15	21	21	15	11	13	16	18	10	12	6	16	3	3	2	9	11	347	99

Figure 1: Distribution of Event Rates by Number of Cells

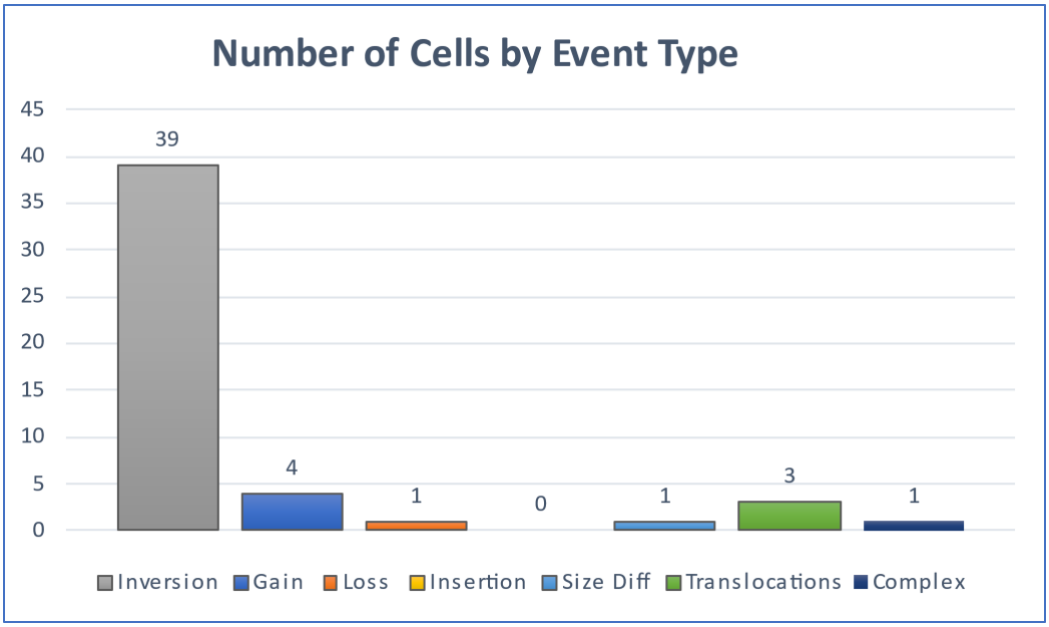


Table 3: Aneuploidy Summary (whole chromosome gain and loss)

	Number of Cells	Percentage of Cells
Gain Event	4	8%
Loss Event	1	2%

Table 4: Aneuploidy detail, by chromosome identity and cell number.

Location	Cell Name	Event	Gain Loss Values
Cell 12 / Chrom-X	Cell 12	Gain	1
Cell 26 / Chrom-X	Cell 26	Gain	1
Cell 34 / Chrom-X	Cell 34	Gain	1
Cell 37 / Chrom-X	Cell 37	Loss	-1
Cell 43 / Chrom-X	Cell 43	Gain	1

Table 5: Inversion and Sister Chromatid Exchange Event Summary

Inversion/SCE Event Counts by Chromosome	P-Arm							Pericentric Inversion	Q-Arm							Grand Total
	Inversion			Exchange					Inversion			Exchange				
	Centric Small	Mid-Arm Mid-Size	Mid-Arm Small	Large Sister Chromatid	Mid-Sized Sister Chromatid	Small Sister Chromatid	Terminal Inversions		Centric Small	Mid-Arm Mid-Size	Mid-Arm Small	Large Sister Chromatid	Mid-Sized Sister Chromatid	Small Sister Chromatid	Terminal Inversions	
1	0	0	6	5	2	2	0	1	1	1	1	5	2	0	0	25
2	1	0	2	6	1	1	0	0	0	0	3	7	3	0	0	24
3	0	0	2	6	2	1	0	1	0	0	1	14	2	1	0	29
4	0	0	3	5	0	2	0	1	0	0	2	8	1	1	0	22
5	0	0	0	4	0	2	0	1	1	1	2	9	3	0	0	22
6	0	0	4	2	0	0	0	0	0	0	7	6	0	0	0	19
7	1	0	2	5	1	0	0	1	0	0	0	6	0	0	0	15
8	0	0	2	4	0	1	0	0	0	0	5	3	3	1	0	19
9	0	0	2	1	0	2	0	0	4	0	6	3	1	2	0	21
10	0	0	1	2	1	0	0	1	0	0	0	8	1	2	0	15
11	0	0	1	3	1	0	0	0	0	0	1	3	1	1	0	11
12	0	0	0	0	0	1	0	0	0	1	4	6	1	0	0	13
13	0	0	0	0	0	0	0	0	0	2	2	11	1	0	0	16
14	0	0	0	0	0	0	0	0	1	0	10	5	0	1	0	17
15	0	0	0	0	0	0	0	0	2	0	2	2	2	2	0	10
16	0	0	2	1	0	0	0	1	0	0	4	4	0	1	0	12
17	0	0	0	0	1	0	0	0	0	0	0	2	2	1	0	6
18	0	0	0	2	1	0	0	1	0	0	7	4	1	1	0	16
19	0	0	0	0	0	0	0	0	1	0	0	1	1	0	0	3
20	0	0	0	0	0	0	0	0	0	0	2	0	1	0	0	3
21	0	0	0	1	0	0	0	0	0	0	0	1	0	0	0	2
22	0	0	0	0	0	0	0	0	0	0	3	0	1	0	0	4
X	0	0	0	2	0	1	0	1	0	0	4	4	0	0	0	11
Y	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
Grand Total	2	0	27	49	10	13	0	9	10	5	66	112	27	14	0	**335**
Inversion Total	2	0	27				0	9	10	5	66				0	101
SCE Total				49	10	13						112	27	14		225

Recurrent SCE/Inversion Event Summary

There are no identified recurrent events in this sample. If recurrent events are identified, graphics of the location(s) are included here.

Notable recurrent events: N/A

Table 6: Insertion Event Summary

There are no insertion events identified in this sample. If insertions are noted, the report contains a table with scored cell number, chromosome ID, location within the chromosome, and color of insert.

Table 7: Translocation Event Summary

Location	Translocation Values	Primary Homolog	Partner(s)	Partner Arms
Cell 16 / Chrom-14	H1(R;R-14p)	H1	R	R-14p
Cell 44 / Chrom-2	H2(8; 8q-2q)	H2	8	8q-2q
Cell 44 / Chrom-8	H2(2; 2q-8q)	H2	2	2q-8q
Cell 50 / Chrom-1	H2(8;8p-1q)	H2	8	8p-1q
Cell 50 / Chrom-8	H2(1;1q-8p)	H2	1	1q-8p

Table 8: Size Difference Summary:

The sample contained one cell with an observed size difference between the homologs of Chr19.

Interpretation / Significance:

Example:
Sample karyotype is diploid with aneuploidy observed in 10% of cells. Aneuploidy observed was gain or loss of a single chromosome homolog. Random SCE and inversion events (events that do not appear repeatedly in over 30% of cells) were seen at the highest prevalence. The sample contained two cells with reciprocal translocations, and one cell with a complex translocation.

Figure 4: Example Karyogram. SCEs present on Chr 2 and Chr 8. Inversion present on Chr 1q (boxed). Inversion Chr Y p-arm (circled).

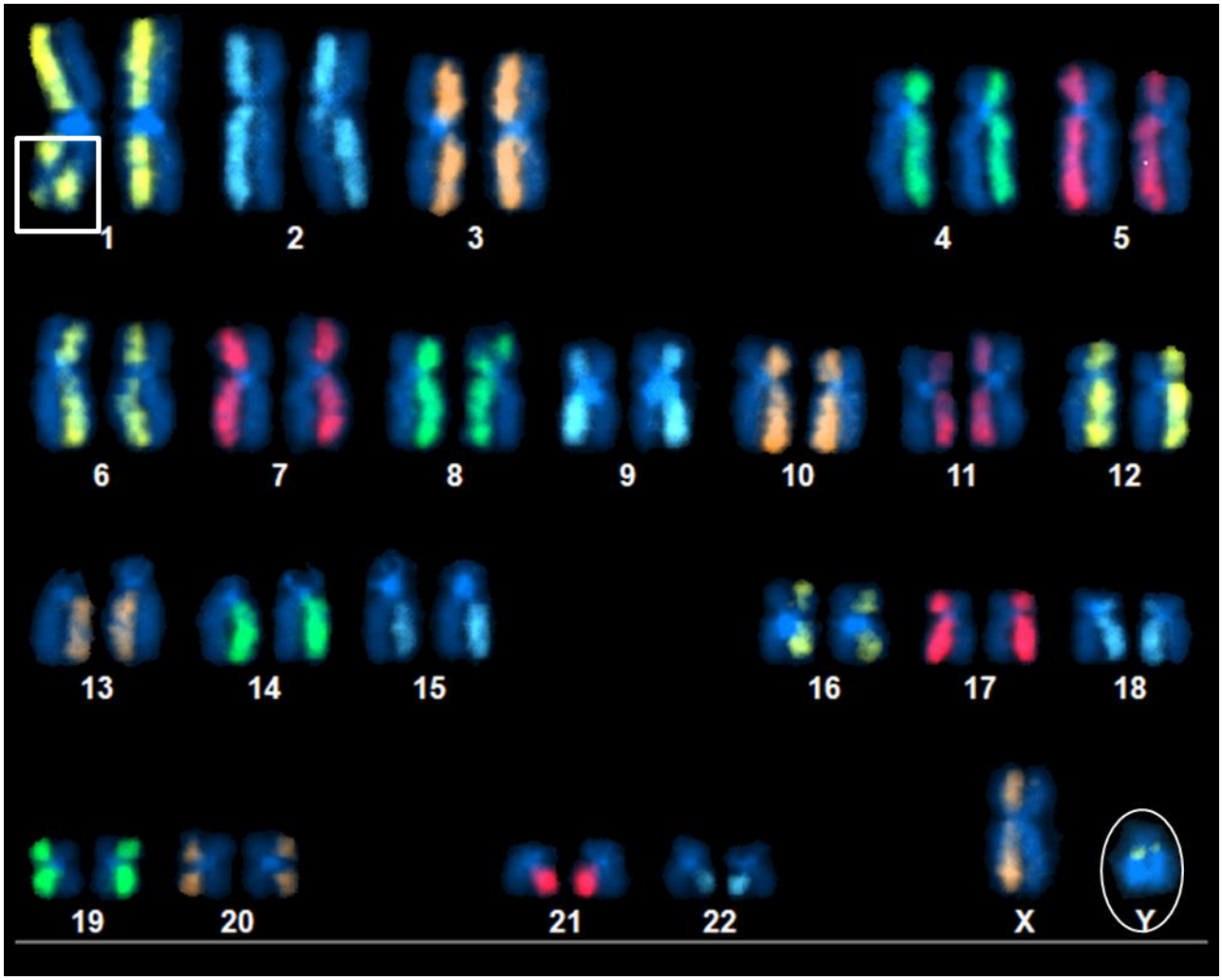
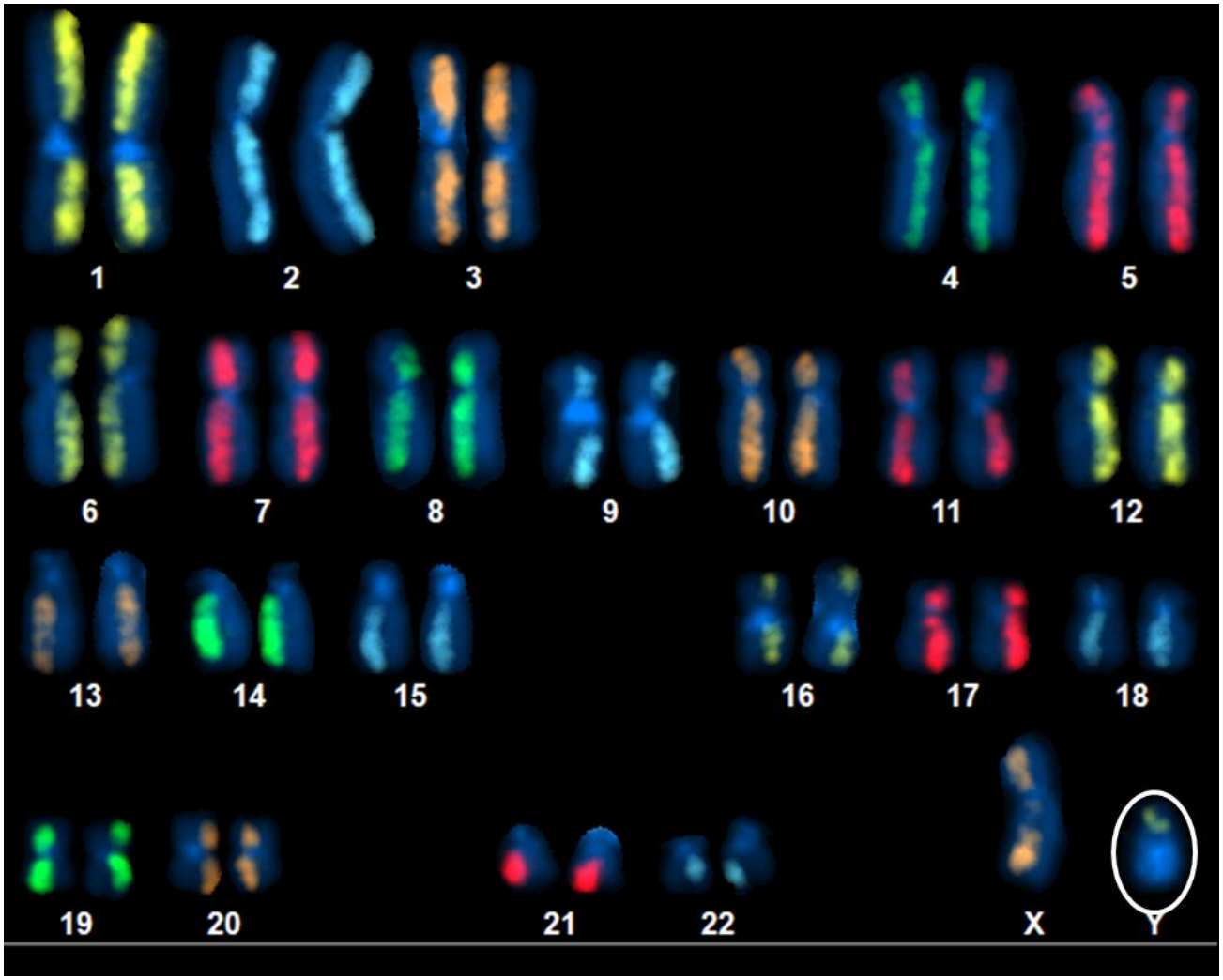


Figure 5: Example karyogram. Structurally normal cell except for inversion Chr Y, p-arm (Circled).



dGH SCREEN 5-Color Whole Genome Assay is for research use only and is not a medical diagnostic test.